



geleophysic dysplasia

Geleophysic dysplasia is an inherited condition that affects many parts of the body. It is characterized by abnormalities involving the bones, joints, heart, and skin.

People with geleophysic dysplasia have short stature with very short hands and feet. Most also develop thickened skin and joint deformities called contractures, both of which significantly limit mobility. Affected individuals usually have a limited range of motion in their fingers, toes, wrists, and elbows. Additionally, contractures in the legs and hips cause many affected people to walk on their toes.

The name of this condition, which comes from the Greek words for happy ("gelios") and nature ("physis"), is derived from the good-natured facial appearance seen in most affected individuals. The distinctive facial features associated with this condition include a round face with full cheeks, a small nose with upturned nostrils, a broad nasal bridge, a thin upper lip, upturned corners of the mouth, and a flat area between the upper lip and the nose (philtrum).

Geleophysic dysplasia is also characterized by heart (cardiac) problems, particularly abnormalities of the cardiac valves. These valves normally control the flow of blood through the heart. In people with geleophysic dysplasia, the cardiac valves thicken, which impedes blood flow and increases blood pressure in the heart. Other heart problems have also been reported in people with geleophysic dysplasia; these include a narrowing of the artery from the heart to the lungs (pulmonary stenosis) and a hole between the two upper chambers of the heart (atrial septal defect).

Other features of geleophysic dysplasia can include an enlarged liver (hepatomegaly) and recurrent respiratory and ear infections. In severe cases, a narrowing of the windpipe (tracheal stenosis) can cause serious breathing problems. As a result of heart and respiratory abnormalities, geleophysic dysplasia is often life-threatening in childhood. However, some affected people have lived into adulthood.

Frequency

Geleophysic dysplasia is a rare disorder whose prevalence is unknown. More than 30 affected individuals have been reported.

Genetic Changes

Geleophysic dysplasia results from mutations in the *ADAMTSL2* gene. This gene provides instructions for making a protein whose function is unclear. The protein is found in the extracellular matrix, which is the intricate lattice of proteins and other molecules that forms in the spaces between cells. Studies suggest that the *ADAMTSL2*

protein may play a role in the microfibrillar network, which is an organized clustering of thread-like filaments (called microfibrils) in the extracellular matrix. This network provides strength and flexibility to tissues throughout the body.

Mutations in the *ADAMTSL2* protein likely change the protein's 3-dimensional structure. Through a process that is poorly understood, *ADAMTSL2* gene mutations alter the microfibrillar network in many different tissues. Impairment of this essential network disrupts the normal functions of cells, which likely contributes to the varied signs and symptoms of geleophysic dysplasia. Researchers are working to determine how mutations in the *ADAMTSL2* gene lead to short stature, heart disease, and the other features of this condition.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- geleophysic dwarfism

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Geleophysic dysplasia 2
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3280054/>

Other Diagnosis and Management Resources

- GeneReview: Geleophysic Dysplasia
<https://www.ncbi.nlm.nih.gov/books/NBK11168>
- MedlinePlus Encyclopedia: Short Stature
<https://medlineplus.gov/ency/article/003271.htm>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>

- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Short Stature
<https://medlineplus.gov/ency/article/003271.htm>
- Health Topic: Growth Disorders
<https://medlineplus.gov/growthdisorders.html>
- Health Topic: Heart Valve Diseases
<https://medlineplus.gov/heartvalvediseases.html>

Genetic and Rare Diseases Information Center

- Geleophysic dwarfism
<https://rarediseases.info.nih.gov/diseases/2449/geleophysic-dwarfism>

Educational Resources

- American Heart Association: Atrial Septal Defect
http://www.heart.org/HEARTORG/Conditions/CongenitalHeartDefects/AboutCongenitalHeartDefects/Atrial-Septal-Defect-ASD_UCM_307021_Article.jsp
- American Heart Association: Pulmonary Stenosis
http://www.heart.org/HEARTORG/Conditions/CongenitalHeartDefects/AboutCongenitalHeartDefects/Pulmonary-Valve-Stenosis_UCM_307034_Article.jsp
- Boston Children's Hospital: Congenital Anomalies of the Esophagus and Trachea
<http://www.childrenshospital.org/conditions-and-treatments/conditions/congenital-anomalies-of-the-esophagus-and-trachea>
- Disease InfoSearch: Geleophysic Dwarfism
<http://www.diseaseinfosearch.org/Geleophysic+Dwarfism/3009>
- Disease InfoSearch: Geleophysic dysplasia 2
<http://www.diseaseinfosearch.org/Geleophysic+dysplasia+2/8455>
- MalaCards: geleophysic dysplasia
http://www.malacards.org/card/geleophysic_dysplasia

- Orphanet: Geleophysic dysplasia
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2623
- Texas Heart Institute: Valve Disease
<http://www.texasheart.org/HIC/Topics/Cond/valvedis.cfm>

Patient Support and Advocacy Resources

- Human Growth Foundation
<http://hgfound.org/>
- Little People of America
<http://www.lpaonline.org/>
- Resource list from the University of Kansas Medical Center: Dwarfism / Short Stature
<http://www.kumc.edu/gec/support/dwarfism.html>
- Resource list from the University of Kansas Medical Center: Heart / Cardiology Conditions
<http://www.kumc.edu/gec/support/conghart.html>
- The MPS Society (UK)
<http://www.mpsociety.org.uk/diseases/related-diseases/geleo-physic-dysplasia/>

GeneReviews

- Geleophysic Dysplasia
<https://www.ncbi.nlm.nih.gov/books/NBK11168>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22skeletal+dysplasias%22+OR+%22geleophysic+dysplasia%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28geleophysic+dysplasia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- GELEOPHYSIC DYSPLASIA 1
<http://omim.org/entry/231050>
- GELEOPHYSIC DYSPLASIA 2
<http://omim.org/entry/614185>

Sources for This Summary

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